IVD gene

isovaleryl-CoA dehydrogenase

Normal Function

The *IVD* gene provides instructions for making an enzyme called isovaleryl-CoA dehydrogenase. This enzyme plays an essential role in processing proteins obtained from the diet. Normally, the body breaks down proteins from food into smaller parts called amino acids. Amino acids can be further processed to provide energy for growth and development. In cells throughout the body, isovaleryl-CoA dehydrogenase is found within specialized structures called mitochondria. Mitochondria convert energy from food to a form that cells can use.

Isovaleryl-CoA dehydrogenase helps process a particular amino acid called leucine. Specifically, this enzyme is responsible for the third step in the breakdown of leucine. This step is a chemical reaction that converts a molecule called isovaleryl-CoA to another molecule, 3-methylcrotonyl-CoA. Additional chemical reactions convert 3-methylcrotonyl-CoA into molecules that are used for energy.

Health Conditions Related to Genetic Changes

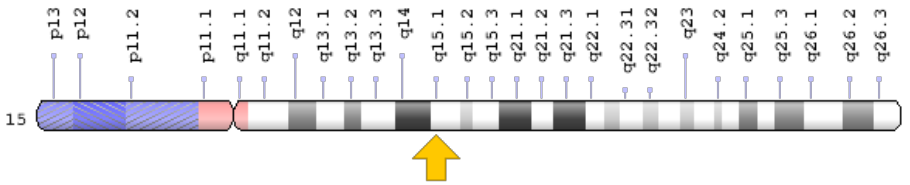
isovaleric acidemia

At least 25 mutations in the *IVD* gene have been identified in people with isovaleric acidemia. Some of these mutations disrupt the normal function of the enzyme, while other mutations prevent the cell from producing any functional enzyme. As a result, the body is unable to break down leucine properly. Defects in leucine processing allow several potentially harmful substances, including a compound called isovaleric acid, to build up to toxic levels in the body. An accumulation of isovaleric acid causes people with isovaleric acidemia to have a characteristic odor of sweaty feet. The buildup of isovaleric acid and related compounds also damages the brain and nervous system, leading to poor feeding, lack of energy (lethargy), seizures, and the other signs and symptoms of isovaleric acidemia.

Chromosomal Location

Cytogenetic Location: 15q15.1, which is the long (q) arm of chromosome 15 at position 15.1

Molecular Location: base pairs 40,405,485 to 40,435,948 on chromosome 15 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ACAD2
- isovaleryl CoA dehydrogenase
- IVD_HUMAN

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): The cause of isovaleric acidemia is a congenital deficiency of isovaleryl-CoA dehydrogenase, which mediates formation of 3-methylcrotonate
  https://www.ncbi.nlm.nih.gov/books/NBK27945/#A3109

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28IVD%5BTIAB%5D%29+AND+%28isovaleric+acidemia%5BTIAB%5D%29%29+OR+%28isovaleryl+Coenzyme+A+dehydrogenase%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- ISOVALERYL-CoA DEHYDROGENASE
  http://omim.org/entry/607036

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=IVD%5Bgene%5D
- HGNC Gene Family: Acyl-CoA dehydrogenase family
  http://www.genenames.org/cgi-bin/genefamilies/set/974
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6186
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3712
- UniProt
  http://www.uniprot.org/uniprot/P26440

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